OCR
RECOGNISING ACHIEVEMENT

ADVANCED GCE

HUMAN BIOLOGY

Genetics, Control and Ageing

F225

Candidates answer on the question paper.

OCR supplied materials:
None

Other materials required:
- Electronic calculator
- Ruler (cm/mm)

Thursday 27 January 2011
Morning

Duration: 1 hour 45 minutes

OCE/26223

F225

Candidate forename		Candidate surname	

Centre number						Candidate number				

INSTRUCTIONS TO CANDIDATES

- Write your name, centre number and candidate number in the boxes above. Please write clearly and in capital letters.
- Use black ink. Pencil may be used for graphs and diagrams only.
- Read each question carefully. Make sure you know what you have to do before starting your answer.
- Write your answer to each question in the space provided. If additional space is required, you should use the lined pages at the end of this booklet. The question number(s) must be clearly shown.
- Answer **all** the questions.
- Do **not** write in the bar codes.

INFORMATION FOR CANDIDATES

- The number of marks is given in brackets **[ ]** at the end of each question or part question.
- The total number of marks for this paper is **100**.
- Where you see this icon you will be awarded marks for the quality of written communication in your answer.
- You may use an electronic calculator.
- You are advised to show all the steps in any calculations.
- This document consists of **28** pages. Any blank pages are indicated.

Answer **all** the questions.

1 A stroke, or cerebrovascular accident (CVA), occurs when the blood supply to part of the brain is disrupted causing brain cells to die.

(a) Explain why a lack of blood flow to part of the brain results in the death of brain cells.

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(b) The Framingham Study is a long-term epidemiological study of the factors which increase the risk of a stroke occurring.

Data from the study has been used to construct the Framingham Stroke Risk Function (FSRF) score. This score is used to assess the risk of a stroke occurring in an individual.

Fig. 1.1 shows four of the risk factors identified in the study and the associated FSRF score.

gender	FSRF score
male	0
female	6

systolic blood pressure (mmHg)	FSRF score
< 120	0
120–139	1
140–159	2
160–179	3
> 179	4

age (years)	FSRF score
55–59	0
60–62	1
63–66	2
67–71	3
72–74	4
75–77	5
78–81	6
82–85	7
86–90	8
91–93	9
> 93	10

diabetes	FSRF score
no	0
yes	6

Fig. 1.1

Fig. 1.2 shows the total FSRF score and the % risk of a stroke occurring within 5 years.

FSRF total score	predicted five year risk of stroke (%)
0–1	5
2–3	6
4	7
5	8
6–7	9
8	11
9	12
10	13
11	14
12	16
13	18
14	19
15	21
16	24
17	26
18	28
19	31
20	34

Fig. 1.2

(i) A diabetic 60-year-old man has a blood pressure of 160 / 90 mmHg.

Use the data in Fig. 1.1 and Fig. 1.2 to predict the risk of this man having a stroke in the next 5 years.

Show your working.

Answer = ... **[2]**

(ii) Explain how high blood pressure can lead to a reduction of blood flow in arteries supplying blood to organs such as the brain.

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(c) Studies have been carried out on the correlation between the percentage risk of a stroke occurring as calculated by an FSRF score, and performance in memory tests.

The study compared groups of participants who had not previously had a stroke.

- One group had a 2% chance of a stroke occurring in the next 10 years.
- The other group had a 20% chance of a stroke occurring in the next 10 years.

Predict which of the two groups would be expected to perform **least** well in memory tests. Give reasons for your prediction.

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(d) Describe the techniques that can be used to improve memory in patients who have suffered a stroke.

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(e) Memory loss is also a symptom of Alzheimer's disease. An investigation was carried out to distinguish between memory loss due to Alzheimer's disease and memory loss due to strokes.

The activity of the brain was monitored during memory tests using positron emission tomography (PET).

Describe the method used in carrying out a PET scan of the brain when investigating memory loss.

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(f) In stroke patients, most activity was recorded in the frontal cortex of the brain during the memory tests. In Alzheimer's patients, most activity was recorded in the area of the brain known as the hippocampus.

Fig. 1.3 is a diagram of the brain. The hippocampus and pons are labelled on the diagram. Other areas of the brain have been labelled **A** to **D**.

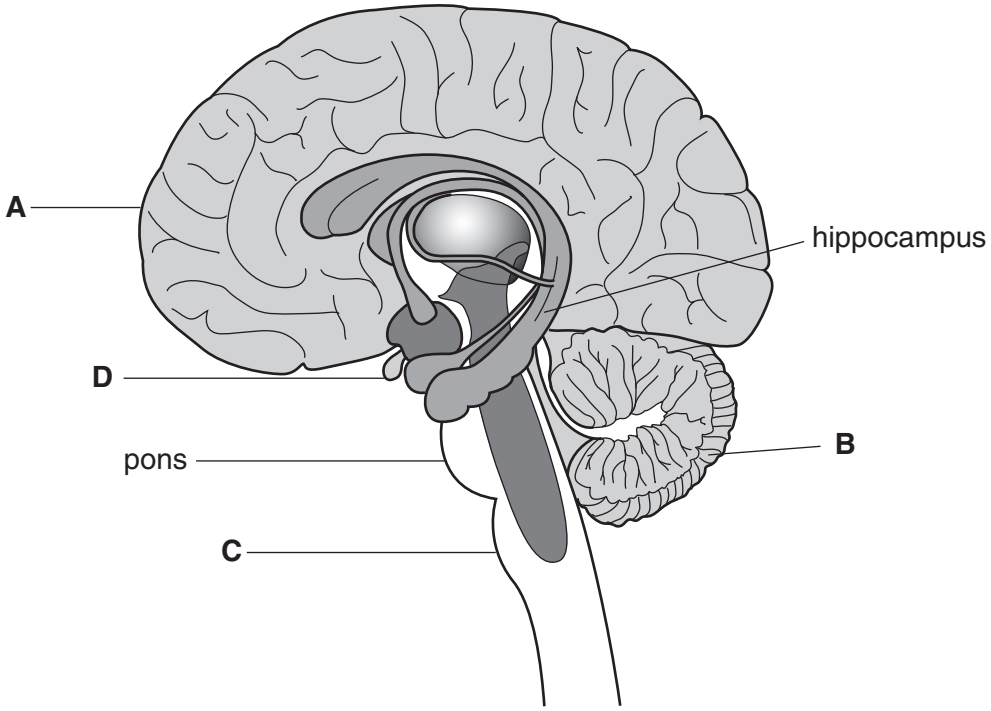

Fig. 1.3

(i) With reference to Fig. 1.3, complete the table by inserting the correct letter for each description.

description of region in brain	letter
frontal cortex	
region responsible for controlling heart rate	
region responsible for controlling blood pressure	

[3]

(ii) Describe the changes in brain tissues that are associated with Alzheimer's disease.

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[Total: 24]

2 Hormone replacement therapy (HRT) is used to relieve menopausal symptoms.

(a) Describe the changes which occur during and after the menopause **and** explain how changes in hormone levels are associated with these changes.

In your answer you should provide a balanced description of both the changes that occur and how the levels of the hormones change.

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(b) Some women use herbal remedies to treat menopausal symptoms.

From 2011, in the United Kingdom, all herbal products sold as medicines will need to be registered with the UK Medicines and Healthcare Products Regulatory Agency.

(i) Suggest what evidence the regulatory agency will be considering before they register a herbal remedy for the treatment of menopausal symptoms.

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..... **[3]**

(ii) Suggest **one** possible active ingredient which may be found in a **herbal** remedy for menopausal symptoms.

..... **[1]**

(c) The use of HRT has been linked to an increased risk of developing some cancers, such as ovarian cancer.

Fig. 2.1 shows the percentage survival at one and five years from diagnosis of **ovarian cancer** in women, in England and Wales from 1971 to 1995.

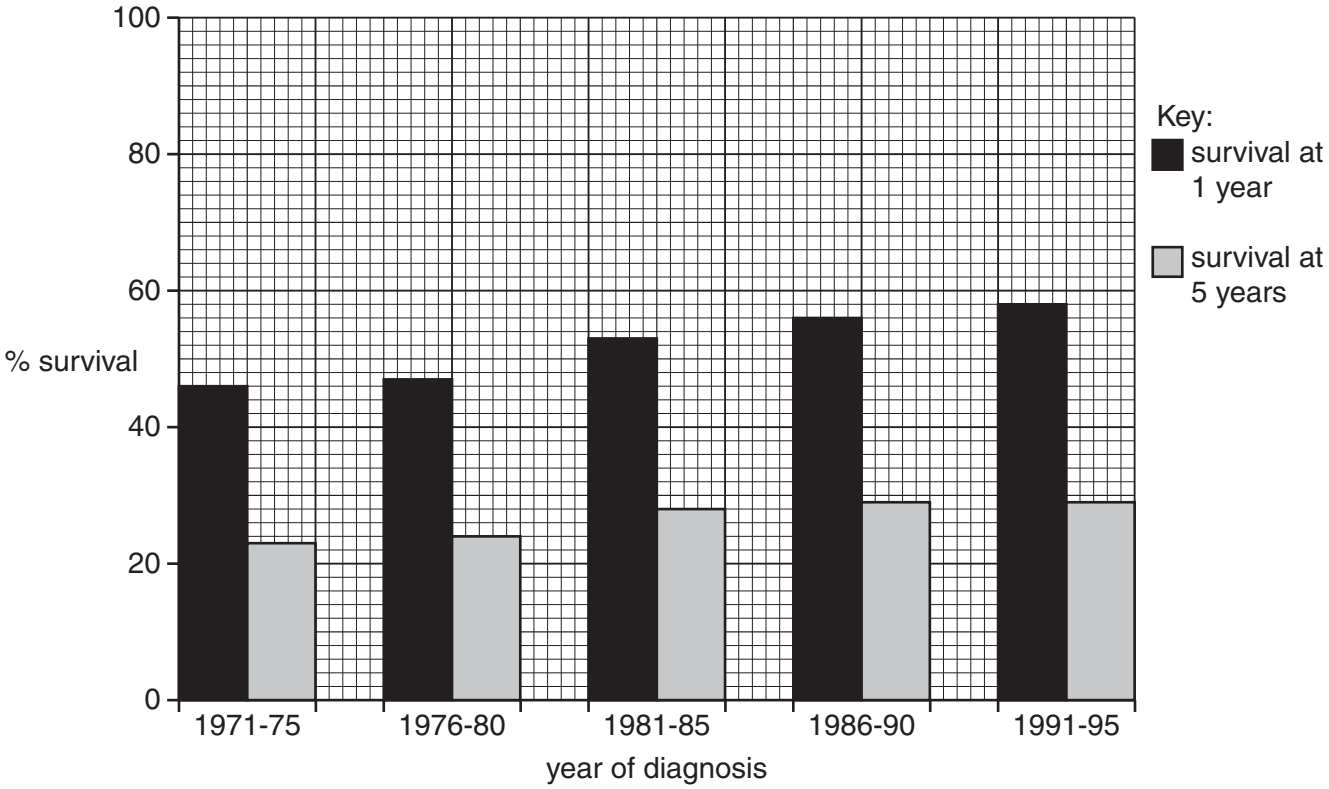

Fig. 2.1

With reference to Fig. 2.1, describe the changes in percentage survival for **ovarian** cancer between 1971 and 1995.

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(d) Fig. 2.2 shows the percentage survival at one and five years from diagnosis of **breast cancer** in women, in England and Wales over the same time period.

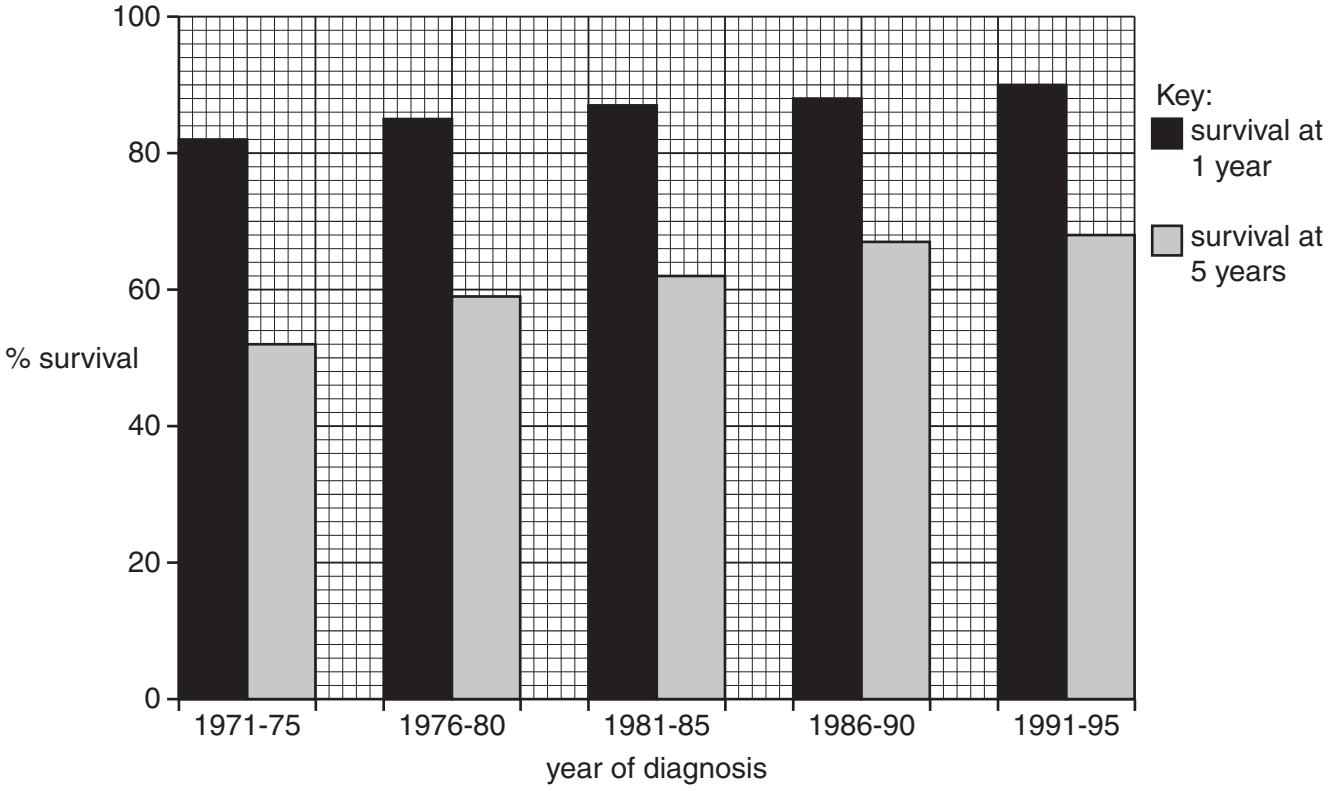

Fig. 2.2

One important factor in percentage survival for cancer is the stage of the disease at the time of diagnosis.

The earlier the stage of the disease on diagnosis, the higher the chance of survival.

(i) Using the data in Fig. 2.1 and Fig. 2.2, describe **two** pieces of evidence which suggest that breast cancer is usually diagnosed earlier than ovarian cancer.

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(ii) Suggest why breast cancer is usually diagnosed earlier than ovarian cancer.

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..... **[2]**

(iii) Suggest **one** method of treating ovarian cancer.

..... **[1]**

[Total: 23]

3 One important function of the kidney nephron is selective reabsorption. This involves the rapid transfer of water across cell surface membranes. The rapid transport of water requires the presence of protein channels known as aquaporins.

Fig. 3.1 is a diagram of a nephron.

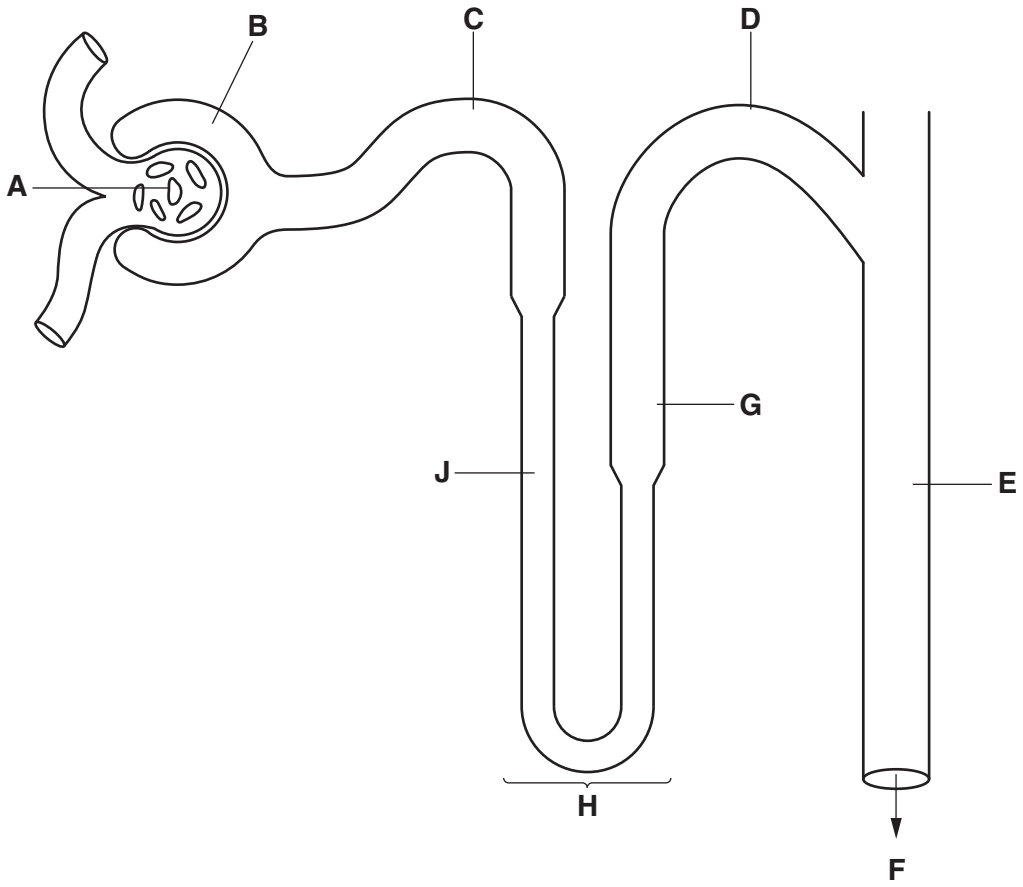

Fig. 3.1

(a) With reference to Fig. 3.1, complete the table by inserting the correct letter for each description.

description of region of nephron	letter
region where numbers of aquaporins vary in tubule wall cells	
region where tubule wall cells are modified to provide filtration slits	
region where aquaporins and glucose transport proteins are present in tubule wall cells	
region where aquaporins are always present in the tubule wall cells but no other transport proteins are present	
region where **no** aquaporins are present in the tubule wall cells	

[5]

(b) The action of antidiuretic hormone (ADH) on cells, such as cells of the nephron, has been investigated using a technique known as confocal laser microscopy.

The technique is carried out as follows:

- cells are mixed with a fluorescent 'marker' which attaches to aquaporins on vesicles in the cytoplasm
- cells are treated with ADH
- cells are examined using a confocal laser microscope
- the position of the aquaporins can be identified from the location of the fluorescent markers.

It was observed that, following treatment with ADH, most of the fluorescent markers were found in the region of the cell surface membrane rather than in the cytoplasm.

In the absence of ADH, most fluorescent markers were observed in the cytoplasm.

Describe the feedback mechanism which leads to the release of ADH **and** explain how the action of ADH on cells enables it to control levels of water in blood plasma.

In this question, you should use the experimental observations described in support of your answer.

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(c) By regulating levels of water in blood plasma, ADH is also involved in the control of the concentration of ions such as sodium.

Hyponatraemia is the term used to describe the situation where the concentration of sodium ions in blood plasma is too **low**.

One of the commonest causes of hyponatraemia is **inappropriate** ADH secretion (IADH).

IADH can be caused by some drugs, including MDMA (ecstasy).

(i) Suggest why IADH can cause hyponatraemia.

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(ii) MDMA has been classified as a class A drug under the 'Misuse of Drugs Act 2005', along with other drugs that modify brain function, such as heroin.

Suggest **two biological** reasons why MDMA was classified as a class A drug.

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..... **[2]**

[Total: 18]

4 Huntington's disease is a genetic disease caused by an autosomal dominant allele. If, at fertilisation, both gametes carry the mutant allele, the resultant embryo will not develop. The homozygous dominant genotype is described as 'lethal'.

In some cases, Huntington's disease symptoms do not appear until an individual is aged 30 or over.

(a) Use a genetic diagram to calculate the probability of a child developing Huntington's disease if both parents begin to show symptoms of the disease.

symbol for normal allele .....

symbol for Huntington's allele .....

parental genotypes ....

gametes ....

children genotypes .....

children phenotypes .....

probability of child developing Huntington's = ... % **[5]**

(b) In situations where symptoms of Huntington's appear in one or both partners and there are children, the family is usually referred to a genetic counsellor. One of the options which will be discussed is whether any **existing** children of the couple should be tested for the presence of the Huntington's allele.

Discuss the **ethical** issues which may arise from the option of testing children for the Huntington's allele if one or both parents show symptoms of the disease.

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(c) The Huntington's allele occurs due to a mutation in a gene on chromosome 4.

- In the normal **allele**, the DNA triplet which codes for the amino acid glutamine is repeated up to 35 times.
- In the Huntington's **allele**, the triplet is repeated more than 120 times.
- A repeat of more than 40 triplets results in Huntington's **disease**.
- The larger the number of repeats, the earlier the onset of the symptoms of Huntington's disease.

The resulting protein has 40 or more additional molecules of the amino acid glutamine in its primary structure. This protein accumulates in neurones.

(i) Suggest why an increase in the number of repeats leads to an earlier onset of the symptoms of Huntington's disease.

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..... **[2]**

(ii) State the precise location in the cell where the additional amino acids would be added to make the final protein.

..... **[1]**

(iii) Describe how these amino acids are joined together.

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(d) The altered protein produced by the Huntington's allele is known as **Htt**.

- **Htt** proteins stick together in neurones forming protein bundles known as neuronal inclusions (NIs).
- The NIs can build up in the axons and dendrites of neurones and can also enter the nucleus.
- In the nucleus, NIs prevents transcription occurring. This results in death of the neurones.

(i) State the difference between an axon and a dendrite.

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(ii) State the position of the nucleus in a neurone.

..... **[1]**

(iii) Suggest why preventing transcription results in death of neurones.

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[Total: 19]

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QUESTION 5 STARTS ON PAGE 22

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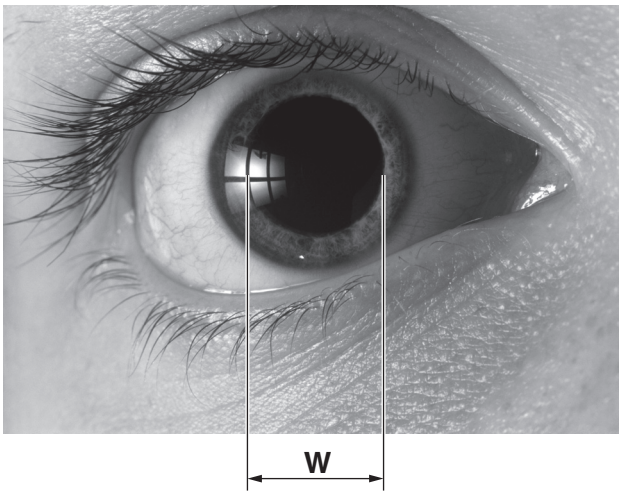
5 Fig. 5.1 is a photograph of a human eye with the pupil fully dilated. The diameter of the pupil is indicated by the line labelled **W**.



magnification x2

Fig. 5.1

(a) In bright light, the diameter of the pupil is at its most narrow. The pupil diameter in bright light is 3 mm.

Using the information provided in Fig. 5.1, calculate the % increase in pupil diameter between the size in bright light and the size when fully dilated.

Answer = ... % **[2]**

(b) Name the white area of the eye visible on Fig. 5.1 **and** state its function.

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..... **[2]**

(c) The table below shows some of the features present in the mammalian eye.

Tick **two** of the features through which light must pass **before** it enters the pupil.

feature	tick
retina	
aqueous humour	
iris	
cornea	
lens	

[2]

[Total: 6]

6 Genetically engineered human proteins are used in the treatment of several medical conditions such as diabetes and kidney failure.

Fig. 6.1 is a summary of the stages in the genetic engineering of a human protein.

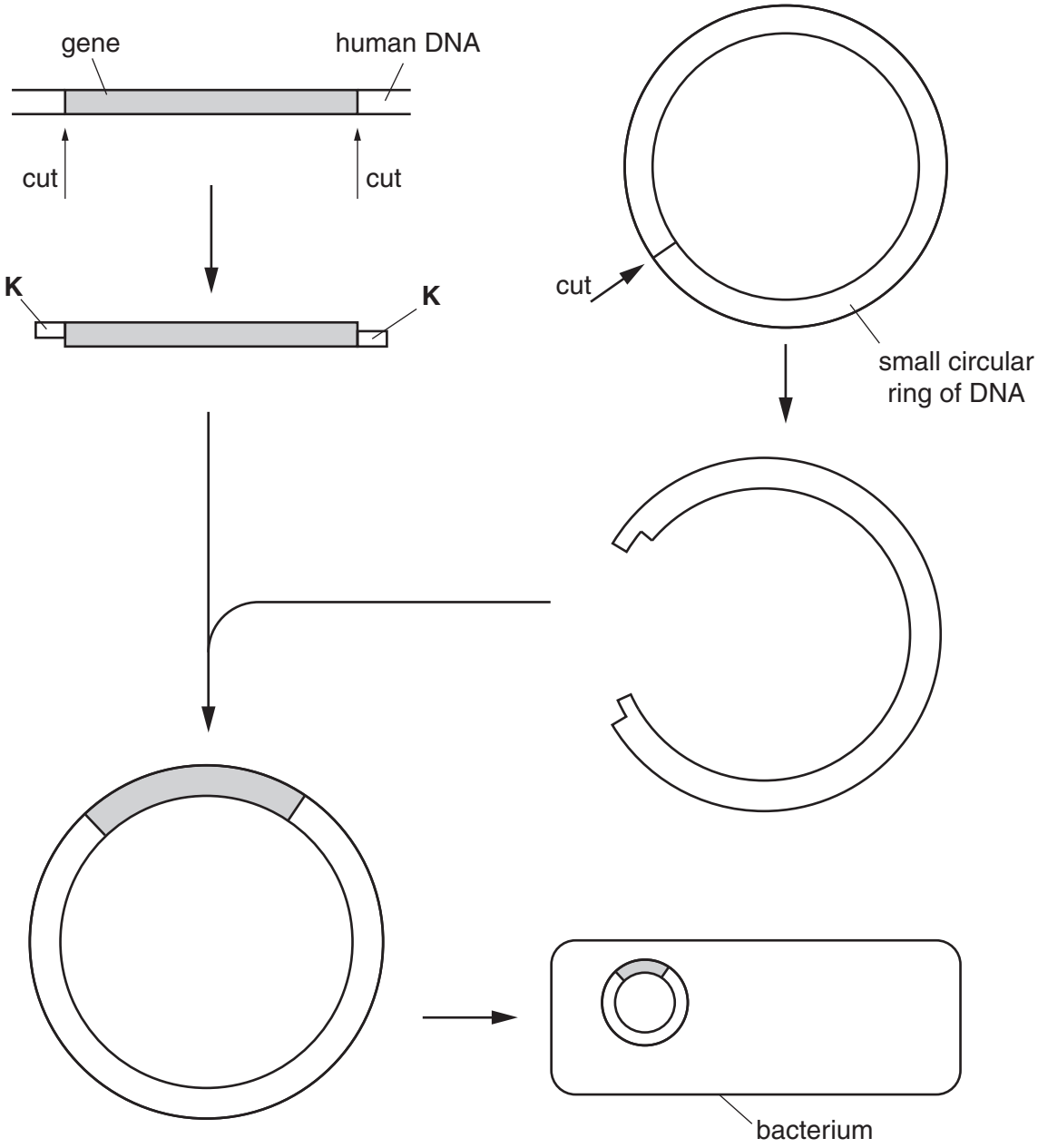

Fig. 6.1

(a) Using the information in Fig. 6.1 and your knowledge, complete the following table:

description	term
the name of the enzyme used to cut DNA	
the name of the feature labelled **K**	
the term for the small circular piece of DNA	
the enzyme used to attach the human gene to the circular DNA	

[4]

(b) Insulin and human growth hormone are two examples of human proteins which can be manufactured using the technique outlined in Fig. 6.1.

The hormone erythropoietin (EPO), used in the treatment of patients on kidney dialysis, is also produced by genetic engineering.

- EPO is a glycoprotein.
- EPO is produced from genetically engineered hamster ovary cells.

(i) State **two structural** differences between the cells used in the genetic engineering of EPO and the bacterial cells used in the genetic engineering of insulin.

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(ii) Suggest why glycoproteins, such as EPO, cannot be produced by genetically engineered bacterial cells.

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(c) Explain why genetically engineered EPO is often given to people on kidney dialysis.

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[Total: 10]

END OF QUESTION PAPER

ADDITIONAL PAGE

If additional space is required, you should use the lined pages below. The question number(s) must be clearly shown.

ADDITIONAL PAGE